Health Care Provider Fact Sheet

Disease Name	**Hemoglobin S-β Thalassemia Disease**
Alternate Name(s)	Beta Thalassemia Sickle Disease
Acronym	Hb β/S
Disease Classification	Hemoglobinopathy Infants whose hemoglobin does not produce enough beta protein have beta thalassemia. It is found in people of Mediterranean descent, such as Italians and Greeks, and is also found in the Arabian Peninsula, Iran, Africa, Southeast Asia and southern China. There are three types of beta thalassemia that also range from mild to severe in their effect on the body. ***Thalassemia Minor* or *Thalassemia Trait***. In this condition, the lack of beta protein is not great enough to cause problems in the normal functioning of the hemoglobin. A person with this condition simply carries the genetic **trait** for thalassemia and will usually experience no health problems other than a possible mild anemia.
Thalassemia Intermedia.	In this condition the lack of beta protein in the hemoglobin is great enough to cause a moderately severe anemia and significant health problems, including bone deformities and enlargement of the spleen. However, there is a wide range in the clinical severity of this condition, and the borderline between thalassemia intermedia and the most severe form, thalassemia major, can be confusing. The deciding factor seems to be the amount of blood transfusions required by the patient. The more dependent the patient is on blood transfusions, the more likely he or she is to be classified as thalassemia major. Generally speaking, patients with thalassemia intermedia need blood transfusions to improve their quality of life, but not in order to survive. ***Thalassemia Major* or *Cooley's Anemia*.** This is the most severe form of beta thalassemia in which the complete lack of beta protein in the hemoglobin causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. These extensive, lifelong blood transfusions lead to iron-overload which must be treated with chelation therapy to prevent early death from organ failure.
Symptoms	Most children with thalassemia major appear healthy at birth, but during the first year or two of life they become pale, listless and fussy, and have a poor appetite. They grow slowly and often develop jaundice (yellowing of the skin).
Natural history without treatment	The spleen, liver, and heart soon become greatly enlarged. Bones become thin and brittle; face bones become distorted, and children with thalassemia often look alike. Heart failure and infection are the leading causes of death among children with untreated thalassemia major. Children with thalassemia intermedia may develop some of the same complications, although in most cases, the course of the disease is mild for the first two decades of life.
Treatment	**Red Blood Cell Transfusion** Because there is no natural way for the body to eliminate iron, the iron in the transfused blood cells builds up in a condition known as "iron overload" and becomes toxic to tissues and organs, particularly the liver and heart. Iron overload typically results in the patient's early death from organ failure. **Chelation Therapy**: To help remove excess iron, patients undergo the difficult and painful infusion of a drug, Desferal. Reduced mortality and morbidity with appropriate penicillin prophylaxis.
Emergency Medical Treatment	See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/StaticContent/ACT/ACT-sheet_Hb_Sbeta_plus_thal_FSA.pdf
Inheritance	Autosomal recessive
General population incidence	1:250,000
Genetests Link	www.geneclinics.org
Support Group	Cooley's Anemia Foundation http://www.cooleysanemia.org/ Sickle Cell Information Center http://www.scinfo.org/ Sickle Cell Disease Association of America, Inc. http://www.sicklecelldisease.org

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